



Need to talk? Call us **0845 2412173** Monday to Friday
9am to 5pm



Supporting those affected by
Inherited Metabolic Disorders

Tyrosinaemia Type 1

Tyrosinaemia Type 1 is a rare metabolic disorder characterised by raised levels of the amino acid, tyrosine in the blood. The disorder occurs when there is an absence or a deficiency of an enzyme known as fumarylacetoacetate hydrolase which is needed to break down tyrosine. If this amino acid is not broken down there is an accumulation of tyrosine breakdown products in the body especially the liver, the kidneys and the brain.

Symptoms vary greatly from case to case. The most common clinical feature is liver failure in young children but it can present later in childhood. Those affected are at a higher risk of liver cell cancer. The kidneys are affected and in some cases the heart too. Tyrosinaemia Type 1 presents during the first few weeks of life. Symptoms can include failure to grow and gain weight, lethargy, irritability, fever, diarrhoea, bloody stools, vomiting, bruising easily, an enlarged liver and jaundice. Sometimes there may be a cabbage-like odour. Children can have repeated neurological crises which can last for days. Symptoms can progress and untreated, this condition is often fatal before age 10. Management for this condition is lifelong.

Synonyms

Alternative names for this condition are:

- Congenital Tyrosinosis
- Fumarylacetoacetase Deficiency
- Hepatorenal Tyrosinaemia
- Hereditary Tyrosinaemia Type 1 / HT-1



Further information about this condition is available from Climb.

Disclaimer

[Please read our disclaimer and information on data protection.](#)

Updated 28/09/17

Telephone: **0845 241 2173**
Email: contact@climb.org.uk

Freephone: **0800 652 3181**
Website: www.climb.org.uk

Children Living with Inherited Metabolic Diseases is a charity registered in England and Wales (1089588) in Scotland (SC044634) and a Company Limited by Guarantee 4267454



ClimbHQ



ClimbHQ



TeamClimb